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ORIGINAL ARTICLE

Determining the Sex Assigned at Birth of Transgender and Nonbinary Populations in Administrative Claims Databases Utilizing Diagnostic and Procedure Codes

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Abstract

Purpose: Widespread conflation of sex assigned at birth and gender has hindered the identification of transgender and nonbinary people in large datasets. The study objective was to develop a method of determining the sex assigned at birth of transgender and nonbinary patients utilizing sex-specific diagnostic and procedural codes, for future use in administrative claims databases, with a goal of expanding the available datasets for exploring sex-specific conditions among transgender and nonbinary people.

Methods: Authors reviewed indexes of International Classification of Diseases (ICD) and Current Procedural Terminology (CPT) codes, and medical record data from a single institution's gender-affirming clinics. Sex-specific ICD and CPT codes were identified through author review and consultation with subject experts. Patient's sex assigned at birth determined by chart review, as a gold standard, was compared with sex assigned at birth determined by querying their electronic health records for natal sex-specific codes.

Results: Sex-specific codes correctly identified 53.5% ($n = 364$) of transgender and nonbinary patients assigned female sex at birth, and 17.3% ($n = 108$) of those assigned male sex at birth. Codes were 95.7% and 98.3% specific for assigned female and male sex at birth, respectively.

Conclusions: ICD and CPT codes can be used to specifically determine the sex assigned at birth in databases where this information is not recorded. This methodology has novel potential for use in exploring sex-specific conditions among transgender and nonbinary patients in administrative claims data.

Keywords: administrative claims data; quantitative method development; sex assigned at birth; sex and gender conflation; transgender and nonbinary

Introduction

Datasets with large sample sizes are important research tools, particularly when studying rare outcomes and minority populations. However, widespread conflation of sex assigned at birth and gender identity has hindered identification of transgender and nonbinary people in large datasets and limited research regarding gender minority populations.[1,2] Sex assigned at birth refers to the declaration of an infant's sex, typically following brief inspection of external genitalia, whereas gender identity refers to one's internal sense of gender. The terms transgender and nonbinary refer to individuals whose gender identity differs from their sex assigned at birth, including those whose gender identity does not conform to the traditional gender binary.[3]

Survey utilization of single-item questions regarding sex or gender, rather than the recommended two-step approach to determine sex assigned at birth and gender identity, results in an assumption of cisgender identity and the inability to identify transgender and nonbinary people.[4] Similar to national surveys, administrative claims databases lack documentation of sex assigned at

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birth and gender identity, and instead list only "legal sex"—or the sex listed on one's insurance record—which is an unreliable indicator of either sex assigned at birth or gender identity.[5]

Transgender and nonbinary people often lack access to competent medical care and face disproportionately high levels of discrimination in health care.[6,7] To address these disparities, there have been calls for greater research of this population. However, use of data from national surveys and administrative databases is hindered by the inability to identify gender minority people, which stifles researchers' ability to disentangle the influence of sex and gender on various health conditions and forces reliance on smaller convenience sample surveys.[5,8]

The dearth of databases with accurate sex and gender information has led some researchers to explore using diagnostic codes to identify transgender and nonbinary people in administrative claims databases. International Classification of Diseases (ICD) codes for gender dysphoria have been used to identify transgender people in administrative claims databases and this identification appears to closely align with clinician notations in electronic health records that describe patients as transgender.[5,8,9] Although these methods have been useful in identifying transgender people in administrative claims datasets, research continues to be limited by lack of information regarding sex assigned at birth.

Many health analyses are stratified by sex—though are often flawed owing to cis-cenric and cis-normative assumptions resulting in a singular and nonspecific question regarding sex and gender. There has been significant interest in understanding differences in health conditions—from depression and anxiety to cardiovascular disease—by both sex and gender, given differences in disease patterns in the general population and hypotheses of the roles of sex and gender in disease pathophysiology and diagnosis.

The ability to identify the sex assigned at birth of transgender and nonbinary people in administrative claims data would significantly expand our ability to explore these questions of sex and gender. For example, one could more reliably compare cardiovascular disease outcomes among transgender and nonbinary people and cisgender individuals assigned female sex at birth (AFAB), or among transgender and nonbinary people AFAB who do and do not have testosterone prescriptions, to better understand the impact of sex, gender, and hormones on cardiovascular outcomes.

To address this challenge and broaden administrative claims data utilization in research on gender minority populations, we developed a pilot method to determine the likely sex assigned at birth of transgender and nonbinary people utilizing natal sex-specific ICD and Current Procedural Terminology (CPT) codes. To our knowledge, determining the sex assigned at birth of transgender and nonbinary people using diagnostic and procedural codes is a novel approach.

Materials and Methods

This study was determined to be exempt by the institution's IRB (HUM00181480).

Developing a list of natal sex-specific ICD and CPT codes

One author (C.H.) manually reviewed all 2015 ICD-9 Diagnosis and Procedure codes and CPT codes using www.icd9data.com (Alkaline Software) and https://coder.aapc.com (American Academy of Professional Coders, Salt Lake City, UT), respectively. Codes that were likely natal sex-specific based on reference to reproductive organs or gametes were identified. The Centers for Medicare and Medicaid Services' General Equivalence Mapping crosswalk was used to convert ICD-9 codes to ICD-10 codes (https://www.cms.gov/Medicare/Coding/ICD10/2018-ICD-10-CM-and-GEMs). Because not all natal sex-specific ICD-10 codes were captured by this mapping, an additional manual review of all 2021 ICD-10 Diagnosis and Procedure Codes was completed using https://www.icd10data.com (Alkaline Software).

Both ICD-9 and 10 codes were captured to allow our coding methodology to be applied to data before the mid-2015 transition to ICD-10.

The likely natal sex-specific codes were classified as specific for individuals AFAB or assigned male sex at birth (AMAB); as discussed in the limitations, this classification lacks the nuance to differentiate individuals with differences of sex development (DSD). Particularly vague codes were excluded (e.g., ICD-10 N94.6: Unspecified condition associated with female genital organs and menstrual cycle).

Codes that refer to anatomy that may develop from gender-affirming hormonal or surgical interventions (e.g., vulva, vagina, breast, penis, and scrotum) were also excluded given that these structures are thus are not specific to sex assigned at birth. Of note, codes

relating to the cervix were included as AFAB specific. Although it is possible these codes could be used for rare AMAB individuals who underwent vaginoplasty with neocervix creation, neocervix creation is not standard of care in the setting of vaginoplasty, and cervical cancer screening following vaginoplasty is not recommended given the lack of native cervical tissue.[10]

Codes were manually reviewed by a second author (H.P.C.), a physician with expertise in gender affirming care, and discrepancies in interpretation of natal sex specificity were resolved through discussion. When consensus on the interpretation of codes' natal sex specificity was unable to be reached, classification was determined by expert subspecialists familiar with code(s) utilization.

Data source, study setting, and participants

Using Microsoft SQL Server Management Studio (Microsoft Corp., Redmond, WA) and the institution's Data Warehouse, all patients who received care at a single academic institution's Department of Obstetrics and Gynecology's dedicated gender-affirming care clinics from January 1, 2015 to December 31, 2019 were identified ($N = 1198$). We used the label "transgender and nonbinary" broadly to this group of patients, although we recognized this as an umbrella term that may not be used by all patients when self-identifying.

In these clinics, all patients were asked their sex assigned at birth, gender identity, and pronouns on intake forms and this was subsequently documented in the electronic health records both in a gender identity form and in provider notes. Manual chart review of clinician notation from gender-affirming care clinic visits was then completed using the institution's electronic health record system (Epic Systems Corp., Verona, WI) to determine each patients' sex assigned at birth; this determination of sex assigned at birth was regarded as our gold standard.

Code methodology

Toad for Oracle (Quest, Aliso Viejo, CA) was used to query each patient's electronic health record in the institution's Data Warehouse for the determined AFAB or AMAB natal sex-specific ICD and CPT codes. All codes in patients' charts were evaluated, whether those codes were used in association with a gender-affirming care clinic visit or any other visit in the health system.

Using these codes, patients were identified as likely AFAB or AMAB. Many patients' sex assigned at birth was unable to be determined using codes. Of note, some ICD-9 codes use the same number as ICD-10 codes for different diagnoses or procedures. To mitigate this, when querying patients' electronic health records, ICD-9 codes were only used to determine the sex assigned at birth for codes given to patients before October 8, 2015 and ICD-10 codes were only used to determine the sex assigned at birth for codes given to patients after October 8, 2015 when the institution transitioned from using the ICD-9 to the ICD-10 coding system.

Any disagreements regarding patients' sex assigned at birth between chart review and code methodology were explored by first verifying transcription of sex assigned at birth from chart review. Second, each code that incorrectly determined patients' sex assigned at birth was reviewed to determine whether the code was not actually natal sex-specific, was incorrectly assigned to the patient (i.e., cervical codes used to code for an anal pap smear), or was indicative of a DSD. From this process, the list of codes was refined by removing codes that were not natal sex-specific.

Final natal sex-specific codes

See Supplementary Data S1 for a spreadsheet containing a complete list of the Final AFAB Codes. See Supplementary Data S2 for a spreadsheet containing a complete list of the Final AMAB Codes. Codes ending in "X" denote a parent code with similar codes falling under its heading—all codes under the parent code were included in the final code list.

Data analyses

The frequency of the likely natal sex-specific codes was calculated to identify the most common codes used to determine the sex assigned at birth of the patients in the study population; codes that identified at least 1% of the AMAB or AFAB population are given in Tables 1 and 2, whereas the full list of codes are provided in the Supplementary Data S1 and S2.

Using gender-affirming care clinician documentation of patient's sex assigned at birth as the gold standard, the sensitivity and specificity of the code methodology to determine the sex assigned at birth of the study population was calculated.[11] Differences between patients whose sex assigned at birth was able to be determined versus patients whose sex assigned at birth was unable to be determined using the code methodology were explored.

Finally, our electronic medical records contained a field labeled "sex," sometimes termed "legal sex," which reflected the sex listed with the patient's

Table 1. Comprehensive List of International Classification of Diseases and Current Procedural Terminology Codes Used to Identify At Least 1% of the Study Population as Assigned Female Sex at Birth

Code type	Code	Code description	Frequency	Patients with code, %
ICD Diagnosis	Z12.4	Encounter for screening for malignant neoplasm of cervix	161	13.44
ICD Diagnosis	N93.9	Abnormal uterine and vaginal bleeding, unspecified	73	6.09
ICD Diagnosis	D28.2	Benign neoplasm of uterine tubes and ligaments	51	4.26
ICD Diagnosis	Z90.710	Acquired absence of both cervix and uterus	45	3.76
ICD Diagnosis	D26.0	Other benign neoplasm of cervix uteri	34	2.84
ICD Diagnosis	N94.6	Dysmenorrhea, unspecified	34	2.84
ICD Diagnosis	D26.1	Other benign neoplasm of corpus uteri	22	1.84
ICD Diagnosis	V76.2	Screening for malignant neoplasms of cervix	17	1.42
ICD Diagnosis	Z30.430	Encounter for insertion of intrauterine contraceptive device	17	1.42
ICD Diagnosis	D26.9	Other benign neoplasm of uterus, unspecified	16	1.34
ICD Diagnosis	N92.6	Irregular menstruation, unspecified	15	1.25
ICD Diagnosis	Z97.5	Presence of (intrauterine) contraceptive device	15	1.25
ICD Diagnosis	N92.0	Excessive and frequent menstruation with regular cycle	14	1.17
ICD Diagnosis	E28.2	Polycystic ovarian syndrome	12	1.00
ICD Procedure	0UT74ZZ	Resection of bilateral fallopian tubes, percutaneous endoscopic approach	13	1.09
ICD Procedure	0UT94ZZ	Resection of uterus, percutaneous endoscopic approach	13	1.09
ICD Procedure	0UT24ZZ	Resection of bilateral ovaries, percutaneous endoscopic approach	12	1.00
CPT	81025	Urine pregnancy test, by visual color comparison methods	177	14.77
CPT	58571	Laparoscopy, surgical, with total hysterectomy, for uterus 250 g or less; with removal of tube(s) and/or ovary(s)	59	4.92
CPT	55980	Intersex surgery; female to male	17	1.42
CPT	58300	Insertion of intrauterine device	17	1.42

CPT, Current Procedural Terminology; ICD, International Classification of Diseases.

insurance company and can be changed by patients presenting updated legal documents or identification to registration. In administrative claims data, this singular field was often used in research and termed either sex or gender, although it was not a reliable indicator of either sex assigned at birth or gender identity. We explored the discrepancies in this indicator, which we will call legal sex, sex assigned at birth through the gold standard of chart review, and coding methodology.

Results

A total of 1198 transgender and nonbinary patients were identified who received care at a single academic institution's Department of Obstetrics and Gynecology's dedicated gender-affirming care clinics from May 2015 to January 2020.

Through our gold standard, manual chart review of physician notes, 637 patients were identified as AFAB and 561 patients were identified as AMAB. Zero patients were unable to be identified as either AFAB or AMAB through chart review. Of note, there was one patient with a DSD who was AMAB but born with a uterus, ovaries, and vagina according to documentation within their electronic health record; this patient was identified as AMAB through chart review because the patient clearly stated they had been AMAB. This patient was not excluded from the data analysis to highlight the limitation of this methodology to accurately determine the sex assigned at birth of people with DSDs.

Tables 1 and 2 provide a list of the most common codes used to identify transgender and nonbinary people as AFAB and AMAB, respectively, in this study population.

In this population, the code methodology had a sensitivity of 53.4% and specificity of 95.7% in determining

Table 2. Comprehensive List of International Classification of Diseases and Current Procedural Terminology Codes Used to Identify At Least 1% of the Study Population as Assigned Male Sex at Birth

Code type	Code	Code description	Frequency	Patients with code, %
ICD Diagnosis	D29.21	Benign neoplasm of right testis	38	3.17
ICD Diagnosis	D29.22	Benign neoplasm of left testis	37	3.09
ICD Diagnosis	Z12.5	Encounter for screening for malignant neoplasm of prostate	20	1.67
ICD Diagnosis	E29.1	Testicular hypofunction	14	1.17
CPT	54520	Orchiectomy, simple (including subcapsular), with or without testicular prosthesis, scrotal or inguinal approach	31	2.59
CPT	55970	Intersex surgery; male to female	20	1.67

Table 3. Sensitivity and Specificity for International Classification of Diseases and Current Procedural Terminology Coding Methodology Identifying Patients Assigned Female Sex at Birth Compared with a Gold Standard of Chart Review Methodology

		Chart review methodology	
		AFAB	Not AFAB
Code methodology	AFAB	340	24
	Not AFAB	297	537
		Sensitivity: 53.38%	Specificity: 95.72%

AFAB, assigned female sex at birth.

the sex assigned at birth of transgender and nonbinary patients who were AFAB (Table 3). The code methodology had a sensitivity of 17.3% and specificity of 98.3% in determining the sex assigned at birth of transgender and nonbinary patients who were AMAB (Table 4). If one was to use only the codes listed in Tables 1 and 2 to attempt to identify sex assigned at birth in the study population, we found these most commonly used codes identified 26.71% of AFAB individuals and 6.18% of AMAB individuals.

The code methodology incorrectly determined the sex assigned at birth of 35 patients: 11 were incorrectly identified as AFAB and 24 were incorrectly identified as AMAB. These incorrectly identified patients were largely a result of codes being erroneously assigned to patients (34 of 35), and rarely a result of patients with DSDs (1 of 35).

Patients whose sex assigned at birth was able to be determined by the code methodology on average had a greater number of total encounters in their electronic health record. Individuals who were correctly identified as AFAB by the code methodology had an average of 12.2 visits documented in their electronic health record, compared with 4.6 visits ($p < 0.01$) among those who were not able to be identified as AFAB. Individuals who were correctly identified as AMAB by the code methodology had an average of 19.5 visits documented in their electronic health record, compared with 6.3 visits ($p < 0.01$) among those who were not able to be identified as AMAB.

Table 4. Sensitivity and Specificity for International Classification of Diseases and Current Procedural Terminology Coding Methodology Identifying Patients Assigned Male Sex at Birth Compared with a Gold Standard of Chart Review Methodology

		Chart review methodology	
		AMAB	Not AMAB
Code methodology	AMAB	97	11
	Not AMAB	464	626
		Sensitivity: 17.29%	Specificity: 98.27%

AMAB, assigned male sex at birth.

In addition, those who were correctly identified as AMAB by the code methodology were on average older than individuals who were not able to be identified as AMAB (42.0 years vs. 32.8 years, $p < 0.01$). Age differences were not appreciated for AFAB individuals identified versus individuals not identified as AFAB by the code methodology (27.9 years vs. 27.8 years, $p = 0.83$).

Among patients whose sex assigned at birth was determined to be AFAB by our gold standard chart review methodology, 19.5% had their legal sex listed as male, whereas 21.4% of those AMAB by chart review had their legal sex listed as female. Among those who were AFAB by chart review, and also classified as AFAB based on our coding methodology, 18.2% would have been classified as male if the legal sex field had instead been used to abstract sex or gender data. Similarly, among those who were AMAB by chart review, and also classified as AMAB based on our coding methodology, 36.4% would have been classified as female if the legal sex field had instead been used.

Discussion

This work proposed a novel method of determining the likely sex assigned at birth of transgender and nonbinary people using natal sex-specific ICD and CPT codes, and demonstrated that this can be accomplished with high specificity.

Unsurprisingly, the results suggested natal sex-specific ICD and CPTs codes have poor sensitivity for determining the sex assigned at birth of the study population. Many diagnostic and procedural codes are not natal sex-specific, concordant with much of medical care. In addition, gender minority people may be less likely to seek or receive sex-specific screening and reproductive health care, which may result in fewer natal sex-specific diagnostic and procedural codes being associated with their electronic health records.[12]

Low sensitivity indicates such coding schemes cannot be relied upon to determine the sex assigned at birth of all people. However, in administrative databases containing millions of patients, these methods have the potential to identify the sex assigned at birth of thousands of transgender and nonbinary patients; this was not previously possible.

With these methods, researchers can use existing coding methodology, using ICD gender dysphoria-related codes, to identify transgender and nonbinary

people in administrative databases,[5,8] and then apply the coding methodology presented here to identify sex assigned at birth. As such, this methodology has the potential to unlock access to natal sex-specific health data on much larger cohorts of transgender and nonbinary populations and expand the utility of these databases for researchers studying gender minority health. Exploring differences in health outcomes among transgender and nonbinary people by sex assigned at birth is critical in understanding health issues with sex disparities in the general population such as depression, anxiety, cardiovascular disease, and diabetes, as well as reproductive health.[6,8,9]

Although piloted in an electronic health record system to allow cross check with gold standard chart review, this coding methodology is not intended for future use in medical record systems; this coding methodology is intended for use in administrative claims data where the legal sex on file with the insurance company is currently the only available marker of sex or gender and is not a reliable indicator of either sex assigned at birth or of gender identity. The inaccuracy of legal sex, in extrapolating to sex assigned at birth for example, is highlighted in our study population where 19.5% of those who were AFAB and 21.4% of those who were AMAB would have had their sex assigned at birth misclassified if legal sex was used as a proxy.

This work has several limitations. First, there are likely important differences between individuals who have and do not have natal sex-specific codes associated with their electronic health record. Unsurprisingly, patients with a greater number of health care encounters had a greater likelihood of their sex assigned at birth being able to be determined by the code methodology. As such, the code methodology may be unable to identify the most vulnerable transgender and nonbinary people who lack access to care.[6,7]

In addition, those who were correctly identified as AMAB by the code methodology were on average older than individuals who were not able to be identified as AMAB. One potential reason for this finding is that there are no sex-specific screening recommendations for AMAB individuals that begin at an early age unlike AFAB individuals who are recommended to undergo regular cervical cancer screening starting at 21 years of age.[13] As such, identifying younger AMAB individuals through the code methodology may be challenging; this is an important consideration for researchers utilizing this methodology when considering their study population.

In terms of generalizability, the study population was also derived from dedicated gender-affirming care clinics within an academic department of obstetrics and gynecology. As such, there is likely greater utilization of obstetrics and gynecology-associated codes, which are natal sex-specific, especially among people AFAB, and may have inflated the relative sensitivity for detecting AFAB compared with other populations.

Researchers using this coding methodology should be careful to avoid generalizing their findings to transgender and nonbinary people who cannot be identified by coding methodology, and would be encouraged to control for available variables, such as age among AMAB individuals, which are likely to influence ability to identify sex assigned at birth.

Second, the medical coding process is subject to errors.[14] Among the study population, the sex assigned at birth for 35 patients (2.9%) were coded incorrectly when compared against a gold standard of chart review. This may be owing to provider or medical coder error and, outside the context of the gender-affirming care clinic data explored here, patients not disclosing their transgender identity.[7,15]

Third, although sex is often perceived as a binary of male and female, with the assumption that external genitalia, chromosomes, reproductive organs, and hormones align people as either male or female, such assumptions overlook the diversity of sex seen in individuals with DSDs or intersex individuals. The scope of conditions that should be included in the identification of DSDs is also debated (e.g., hypospadias), further complicating potential inclusion of sex diversity in methodology such as that proposed here. This pilot method of determining likely sex assigned at birth relies heavily on coding related to reproductive organs, and thus does not have the nuance necessary to accurately determine the sex assigned at birth of individuals with DSDs as demonstrated by the patient in the study population.

Conclusion

In conclusion, our findings suggest ICD and CPT codes can be used with high specificity to determine the sex assigned at birth of transgender and nonbinary people.

At present, researchers lack access to accurate sex and gender data in national surveys and administrative claims databases. The ability to extrapolate sex assigned at birth through diagnostic and procedural codes provides a novel opportunity to begin to study natal sex-specific conditions among transgender and nonbinary people in large, previously unusable

databases. Although sex and gender data would ideally be patient reported and verified,[4] we cannot wait for such data to exist to begin to address important health issues for transgender and nonbinary populations. This methodology provides a potential window into health outcomes we hope will be more thoroughly explored as documentation of sex and gender improves.

Authors' Contributions

C.H. performed the chart review, initial coding, data analysis and initial article preparation. A.T. performed the data analysis and provided article revisions. D.S., M.B.M., and V.K.D. aided in conceptualization of the study design, interpretation of the data, and revisions of the article. H.P.C. conceived of the study concept and initial design, reviewed coding, performed data analysis, and provided article revisions.

Author Disclosure Statement

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Supplementary Material

Supplementary Data S1
Supplementary Data S2

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Abbreviations Used

AFAB = assigned female sex at birth
AMAB = assigned male sex at birth
CPT = Current Procedural Terminology
DSD = differences of sex development
ICD = International Classification of Diseases